A case of Wild-type transthyretin amyloid cardiomyopathy represented as a dilated cardiomyopathy

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Introduction: Transthyretin amyloid cardiomyopathy(ATTR-CM) is an under-recognized cause of heart failure in older adults, caused by deposition of transthyretin amyloid fibrils in myocardium inducing thick cardiac wall. Cardiac amyloidosis can appear as a hypertrophic cardiomyopathy and few patients referred for surgical myomectomy are diagnosed with ATTR-CM histologically. The most important diagnostic criteria of cardiac amyloidosis is thick LV wall. It is one of causes of heart failure with preserved ejection fraction. Here, we present a case of 79 year old woman with heart failure who showed dilated cardiomyopathy and was finally diagnosed as ATTR-CM.

Case presentation: A 79-year old Asian female was presented with aggravating NYHA III dyspnea for 3 months. Cardiac exams were conducted including NT-pro BNP(BNP) lab, ECG and echocardiography(TTE). Initial BNP was 1469, ECG showed LBBB and TTE result showed thinning of cardiac walls(especially basal and inferior septum) and severely reduced LV systolic function(EF=20%), consistent with dilated cardiomyopathy. Under suspicion of sarcoidosis, further cardiac evaluations including CAG, MRI and endomyocardial biopsy(EMB) were conducted. CAG showed nonspecific findings and MRI result was consistent with non-ischemic DCM. Contrary to our expectations, EMB result was consistent with amyloidosis. Further evaluations including PYP bone scan, protein/immunofixation electrophoresis and TTG mutation study were conducted. PYP bone scan showed results suspected for amyloidosis. Both PEP/IFE results and TTG mutation analysis showed negative results, indicating wild type ATTR-CM. Patient was treated with standard medications of heart failure. Follow up TTE showed improvement(EF=33%). Tafamidis was added for delaying disease progression. 1-month follow up laboratory finding showed decreased BNP to 844.

Discussion: Typical TTE representation of ATTR-CM includes septal wall thickness and apical sparing compared with mid and basal regions. However, amyloid cardiomyopathy can also be represented as a dilated cardiomyopathy and should be kept in mind as a differential diagnosis until EMB result is inconsistent with amyloidosis.

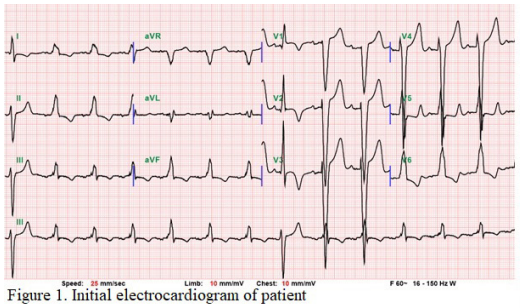


Figure 1. Initial electrocardiogram of patient

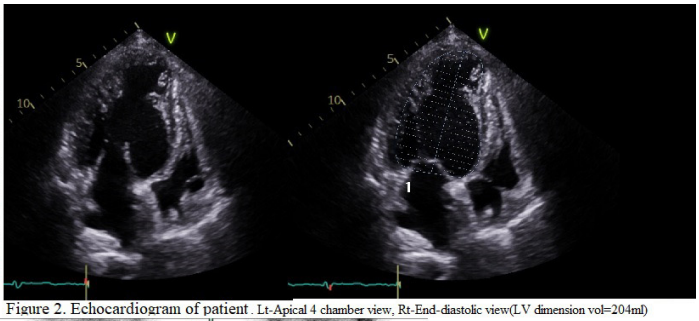


Figure 2. Echocardiogram of patient. Lt-Apical 4 chamber view, Rt-End-diastolic view(LV dimension vol=204ml)

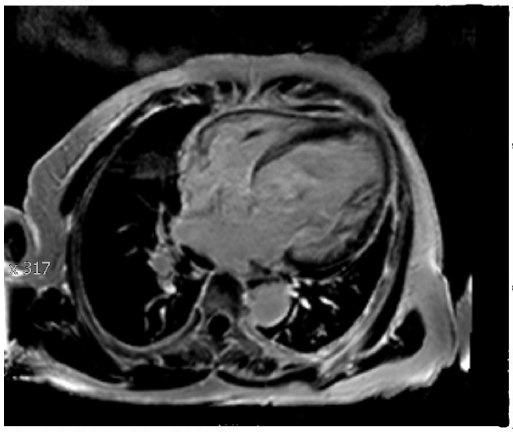


Figure 3. Cardiac MRI of patient.

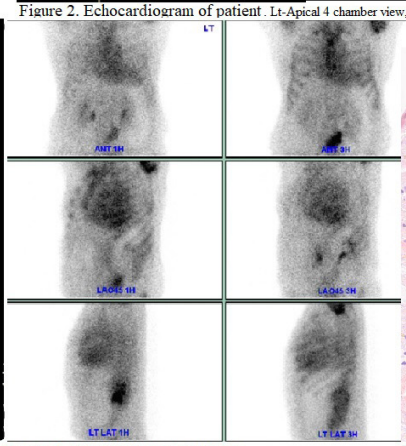
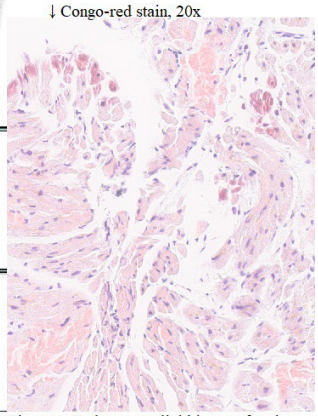


Figure 4. PYP bone scan of patient



↓ Congo-red stain, 20x

Figure 5. Endomyocardial biopsy of patient